Analysis of NIDCR's Rare Disease Portfolio

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Abstract

In order to understand the landscape of rare diseases affecting the dental, oral, and craniofacial skeleton, an analysis was conducted of NIDCR's research grants in FY14. Using manually coded descriptors of NIDCR's funded research based on reading the applications abstracts, a broad categorization was conducted. The aim was to gain an understanding of the current portfolio with regard to the disease research, and to explore needs, gaps, and opportunities in this area. Using analytical tools, additional examination of the research outputs, key players, and the impacts of these outputs are presented. By identifying the hurdles, successes, and state of progress towards clinical readiness in rare diseases, the NIH will be better poised to leverage resources for funding rare disease research in the area of dental, oral, and craniofacial science. The ultimate goal is to look at scientific areas where rare disease research has helped advance knowledge in related, but more common, diseases and pathways.

What are "Rare" diseases?

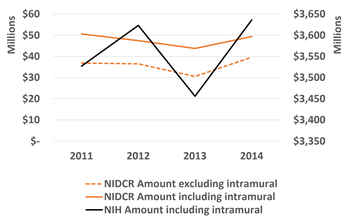
- Diseases which are characterized by a low prevalence (<200,000 people) in the population. They frequently are associated with problems in diagnosis and treatment.
- But, having a rare disease is not so rare. Of the roughly 6,800 rare diseases that are known, about 10% of the US population has some form of rare disease.
- There are more than 5,000 rare disorders that, taken together, affect approximately 20 million Americans.
- One in every 12 individuals in this country has received a diagnosis of a rare disease (from National Organization for Rare Disorders).

Data Sources for FY14 projects list

- RePORT/RCDC- publicly reported "rare diseases" category
- SCORE- NIDCR coding system, pre-defined disease subset
- 121 overlap, 22 in RCDC alone, 113 in SCORE alone

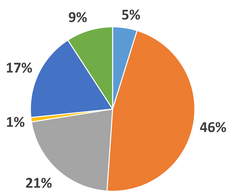
Consolidated list of 256 unique projects used for the analysis
SCORE coding allowed for additional grants to be included with those from RCDC "rare disease" category

NIDCR funding for rare diseases research has remained stable



Mechanism	Project count
Research Project Grant	163
Fellowship	23
Career Development	18
Program Project	17
Intramural	14
Cooperative Agreement	13
NIH Director's Award	6
Grand Total	**254**

Funded research is heavy in basic sciences, with few pre-clinical studies

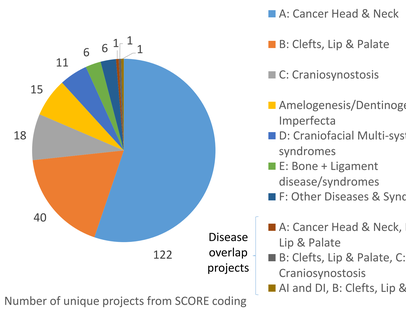


Based on program codes which approximates unique topic areas

- Clinical
- Integrative & Infectious
- Genomics
- Behavioral
- Training
- unassigned

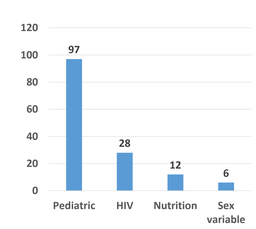
Translational Pipeline	
Clinical Trials	0
Human Subjects	41
Animal + Human subjects	49
Animal studies	117
In vitro	18

More than half the projects are related to head and neck cancer

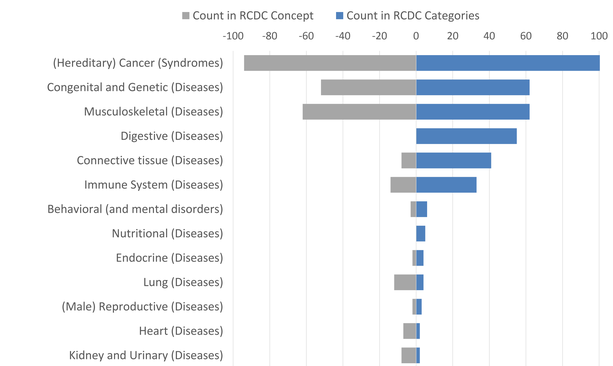


Number of unique projects from SCORE coding

Rare disease research in major policy areas



Disease categorization by ORDR framework



Disease categories from Genetic and Rare Diseases Information Center (GARD)

Publications

- 559 from Jan 2014 to present

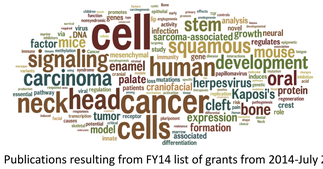
Additional or continued funding of work

- 85 competing renewals
- 170 ongoing projects

Patents/Inventions/Therapeutics

- No records in iEdison and ExPORTER
- iREPORT showed that success stories were mostly in basic sciences

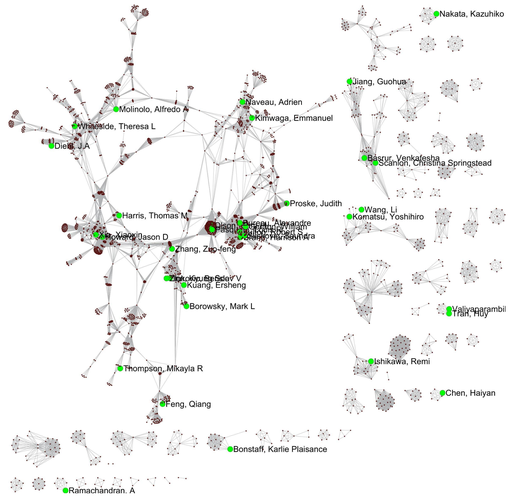
Major themes from research publications



Publications resulting from FY14 list of grants from 2014-July 2015

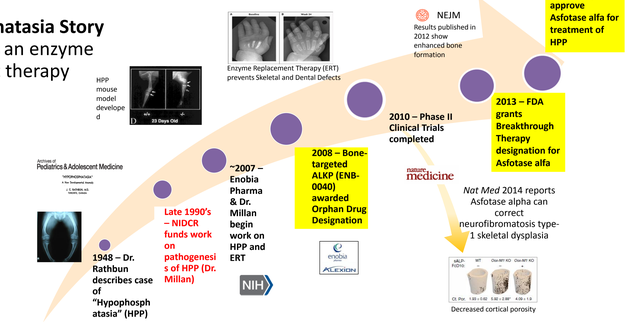
Funded investigators collaborate on rare disease projects

(iClean beta from OPA & Sci2)



The Hypophosphatasia Story

Asfotase alfa as an enzyme replacement therapy



Summary

- Funded research is heavy in basic sciences, with few pre-clinical studies.
- There are no clinical trials in NIDCR portfolio.
- More than half the projects are related to head and neck cancer.

Future Analyses

- Are there other rare disease areas we should be funding?
- What are the hurdles in advancing rare disease research?
- Has rare disease research helped advance discoveries or treatment of more prevalent diseases?